4 General

4.1 Basics

The basic characteristics of a terminology influence its utility and appropriateness in clinical applications. Terminologies should be evaluated within the context of their stated scope and purpose and are intended to complement and utilize those notions already identified by other national and international standards bodies.

This Technical Specification explicitly refers only to terminologies that are primarily designed to be used for clinical concept representation or to the aspect of a terminology designed to be used for clinical concept representation. This Technical Specification will also provide terminology developers and authors with the quality guidelines needed to construct useful and maintainable controlled health terminologies. These tenets do not attempt to specify all the richness which can be incorporated into a health terminology. However, this Technical Specification does specify the minimal requirements, which, if not adhered to, will assure that the terminology will have only limited generalizability and will be very difficult, if not impossible, to maintain. Terminologies which do not currently meet these criteria, can be in compliance with this Technical Specification by putting in place mechanisms to move toward these goals. Principles for implementation are specified in Annex B.

This Technical Specification will provide terminology developers with a sturdy starting point for the development of controlled health terminologies. This foundation serves as the basis from which terminology developers will build robust, large-scale, reliable and maintainable terminologies.

4.2 Concept orientation

The basic unit of a terminology shall be a concept, which is the embodiment of some specific meaning and not a code or character string. Identifiers of a concept shall correspond to one and only one meaning and, in a well-ordered terminology, only one concept may have that same meaning, as specified in ISO 860. However, multiple terms (linguistic representations) may have the same meaning if they are explicit representations of the same concept. This implies non-redundancy, non-ambiguity, non-vagueness and internal consistency.

4.2.1 Non-redundancy

Terminologies shall be internally normalized. There shall not be more than one concept identifier in the terminology with the same meaning, as specified in ISO 704 and E-1284. This does not exclude synonymy, rather it requires that this be explicitly represented.

4.2.2 Non-ambiguity

No concept identifier should have more than one meaning. However, an entry term can point to more than one concept.

EXAMPLE MI as myocardial infarction and mitral insufficiency.

NOTE Some authors have referred to entry terms as an interface terminology.

4.2.3 Non-vagueness

Concept names shall be context free.

EXAMPLE “Diabetes mellitus” should not have the child concept “well controlled”, instead the child concept's name should be “diabetes mellitus, well controlled”.

NOTE Some authors have referred to context free as context laden.

4.2.4 Internal consistency

Relationships between concepts should be uniform across parallel domains within the terminology.

EXAMPLE If heart valve structures are specified anatomically, the diagnosis related to each structure is also specified using the same relationships.

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4.3 Purpose and scope

Any controlled terminology shall have its purpose and scope clearly stated in operational terms so that its fitness for particular purposes can be assessed and evaluated. Where appropriate, it may be useful to illustrate the scope by examples or “use cases” as in database models and other specification tools. Criteria, such as coverage and comprehensiveness, can only be judged relative to the intended use and scope.

EXAMPLE A terminology might be comprehensive and detailed enough for general practice with respect to cardiovascular signs, symptoms and disorders, but inadequate to a specialist cardiology or cardiothoracic surgery unit. Conversely, a terminology sufficiently detailed to cope with cardiology and cardiothoracic surgery might be totally impractical in general practice.

4.3.1 Coverage

Each segment of the healthcare process shall have explicit in-depth coverage, and not rely on broad leaf node categories that place specific clinical concepts together. The extent to which the depth of coverage is incomplete shall be explicitly specified for each domain (scope) and purpose as indicated in 4.3.[9]

EXAMPLE It is often important to distinguish specific diagnosis from categories presently labelled “not elsewhere classified” (NEC), or to differentiate disease severity such as indolent prostate cancer from widely metastatic disease.

4.3.2 Comprehensiveness

The extent to which the degree of comprehensiveness is incomplete shall be explicitly specified for each domain (scope), and purpose as indicated in 4.3. Within the scope and purpose, all aspects of the healthcare process shall be addressed for all related disciplines, such as physical findings, risk factors, or functional status — across the breadth of medicine, surgery, nursing and dentistry. This criterion applies because decision support, risk adjustment, outcomes research and useful guidelines require more than diagnoses and procedures.

EXAMPLES The existing Agency for Healthcare Research and Quality Guidelines, and the Healthcare Finance Administration (HCFA) mortality model.[10]

4.4 Mapping

Government and payers mandate the form and classification schema for much clinical data exchange. Thus, comprehensive and detailed representations of patient data within computer-based patient records should have the ability to be mapped to those classifications, such as ICD-9. This need for multiple granularities is required for clinical healthcare, as well as is specified in ISO/IEC/TR 9789. The degree to which the terminology is mappable to other classifications shall be explicitly stated.[11]

EXAMPLE An endocrinologist may specify more detail about a patient’s diabetes mellitus than a generalist working in a primary care setting, even though both specialities may be caring for the same patient.

4.5 Systematic definitions

In order for users of the terminology to be certain that the meaning that they assign to concepts is identical to the meaning which the authors of the terminology have assigned to these definitions will need to be explicit and available to the users. Further, as relationships are built into terminologies, multiple authors will need these definitions to ensure consistency in authorship.

EXAMPLE The clinical concept “hypertension” might be defined as a consistently elevated blood pressure and needs to be distinguished from a single “BP > 140/85”.

4.6 Formal definitions

A compositional system should contain formal definitions for non-atomic concepts and formal rules for inferring subsumption from the definitions, as specified in E-1712.

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